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Case report

Association between juvenile idiopathic arthritis and osteogenesis imperfecta - case report

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ABSTRACT

The authors report a rare association case of juvenile idiopathic arthritis (JIA) and osteogenesis imperfecta (OI) in a 53 years-old female patient, present a literature review and discuss the radiological aspects of the temporo-mandibular joint involvement. To our knowledge, this is the first case report of JIA an OI association.

Artrite idiopática juvenil e *osteogenesis imperfecta* – relato de caso

Palavras-chave:
Artrite idiopática juvenil
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RESUMO

Os autores relatam o caso de uma paciente de 53 anos que apresenta uma rara associação entre artrite idiopática juvenil (AIJ) e osteogenesis imperfecta (OI), com acometimento poliarticular, incluindo a articulação temporomandibular. Apresentam uma revisão da literatura e uma discussão dos aspectos radiológicos do acometimento da referida articulação. Não foram encontrados relatos de casos com semelhante associação de doenças na literatura especializada.

Introduction

Juvenile idiopathic arthritis (JIA) is the most common chronic rheumatic disease in childhood, and can affect any young individual before the age of 16 years. Its etiology is still unknown, but its physiopathology consists of the chronic inflammation of the synovial membrane of one or more joints. The disease classification is based on its clinical manifestations during the first six months. The polyarticular subtype with positive rheumatoid factor, despite representing less than 10% of all cases, is the most prevalent form in early adolescence. Temporomandibular joint (TMJ) involvement in JIA is quite variable, and can occur in 17% to 87% of patients.[1]

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Osteogenesis imperfecta (OI) is an autosomal dominant hereditary disease defined by bone frailty due to abnormal synthesis of type 1 collagen in the bone matrix. It affects the entire skeleton, predisposing the patient to frequent nontraumatic fractures, causing pain, skeletal deformity, and disability.[2] There may be articular manifestations, such as arthralgia and deformities secondary to fractures, although it does not develop into morning stiffness and bone erosion, as in JIA.

The authors report an unusual association between OI and JIA with TMJ involvement, which is rare in OI, and led the investigation to a retrospective diagnosis of chronic arthritis.

Case report

A female patient, 53 years old, born and raised in the city of Rio de Janeiro, Brazil, came to the Rheumatology Service of the Hospital Universitário Clementino Fraga Filho of the Universidade Federal do Rio de Janeiro (HUCFF/UFRJ) for the treatment of osteoporosis associated with a history of recurrent fractures. Clinical examination revealed blue sclera (Fig. 1A), important limitation in the opening of the mouth (Fig. 1B), and severe deformities of hands and feet (Figs. 2A and 2B). Hormonal investigations discarded osteometabolic diseases, such as hypophosphatasia and tumors of the adrenal cortex. The clinical-epidemiological, laboratory, and radiological findings allowed the diagnoses of congenital syphilis, osteopetrosis, idiopathic juvenile osteoporosis, celiac disease, fibrous dysplasia, and bone tumors to be discarded.

Radiographs of the thoracic spine and pelvis showed marked decrease in bone mass with collapse of several dorsal vertebrae, and bone densitometry showed T-scores of -4.7 SD in the lumbar spine (L1-L4) and -4.6 SD in the total femur. A review of the patient's medical history demonstrated that since age 2 years she had suffered spontaneous fractures, and that her brother had a similar history of fractures. Neither had been diagnosed in the past with regard to the fractures.

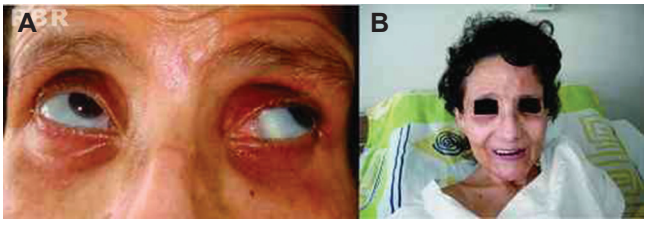

Fig. 1 – A, Blueish sclera in patient with osteogenesis imperfecta associated with juvenile idiopathic arthritis. B, Mouth opening limitation due to temporomandibular joint involvement.

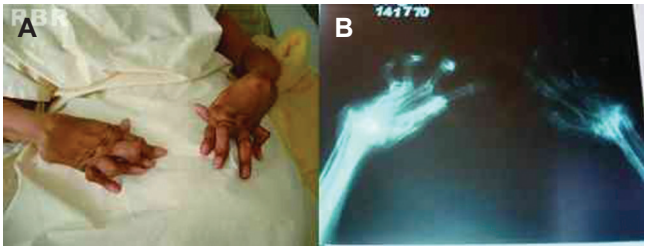

Fig. 2 – A, Hand deformities. B, Corresponding X-ray.

Additionally, the patient had a picture of cumulative symmetric polyarthritis of large and small joints, which started in childhood, associated with sporadic fever and morning stiffness; polyarticular onset JIA was diagnosed at age 15 years. The evaluations at the time of diagnosis showed elevated inflammatory markers (ESR = 72 mm/h), presence of rheumatoid factor (not quantified, but confirmed on other occasions), and irregular use of salicylates and gold salts during periods of arthritis worsening. No other autoantibody was detected.

The diagnosis of JIA associated with OI was clinically confirmed, despite the lack of molecular evidence, and treatment was initiated with an anti-reabsorptive drug (alendronate).

Discussion

JIA was recently classified by the International League of Associations for Rheumatology (ILAR) into seven subtypes; females are more affected by all of them.[3] Polyarticular JIA with positive rheumatoid factor is the form that resembles the adult disease, evolves with severe joint damage and, in general, accompanies the patient throughout life, with periods of exacerbation and inactivity.[4]

JIA patients often have poor oral occlusion due to the effect of the disease on the TMJ.[5] The involvement of the TMJ in JIA is not uncommon and can even occur alone and insidiously. The incidence varies from 17% to 87% of patients, depending on the JIA subtype studied. This condition continues to be one of the most underdiagnosed and undertreated in JIA.[1]

Patients with TMJ involvement may be asymptomatic or have morning stiffness, reduced capacity of mouth opening, rales, trismus, and pain on joint palpation.[1] Patients with more marked alterations are those with longer duration of disease, earlier onset, or those with polyarticular or systemic onset disease.[1]

The TMJ can be investigated by some complementary tests, such as orthopantomogram, ultrasound, computed tomography (CI), or magnetic resonance imaging (MRI).[2] Ultrasonography, an examination that yields good joint assessment, depends largely on the experience of the radiologist. The orthopantomogram is an affordable and low cost-examination, but has the disadvantage of not differentiating chronic from active lesions.

Computed tomography results in better visualization of the mandibular condyle, but does not detect soft tissue alterations that indicate joint inflammation. MRI is regarded as the gold standard for diagnosis, since it is able to demonstrate synovial effusion or proliferation (pannus),[1] as in the reported case.

OI has a higher prevalence in white women. Most patients have blue sclera and family history of the disease. Its classification was described in 1978[6] and reviewed in 2010,[7] but type 1 remains the mildest and most prevalent form, with a prognosis compatible with life and allowing for ambulation.[6] Although there was no molecular verification, this patient likely has type 1, in which the deformities are uncommon and there may not be final height impairment. The alterations observed in extremities are characteristic of JIA without adequate treatment (Figs. 2A and 2B). Additionally, deafness is observed in only 30% of cases.[8]

OI patients can have normal teeth, with a moderate change in color, or quite abnormal dentition. Dental fractures can occur easily and require extractions.[9] The normal dentin is rich in collagen type 1. JIA patients, however, have significantly reduced alveolar bone density.[10] These factors together are enough to interfere with the patient's dental assessment.

During the course of OI, it is possible to observe arthralgia, joint hypermobility, and tendon rupture.[11] The disease itself can lead to deformities in the hands, characterized by swan neck fingers and reversible contractures. This process is attributed to fractures and ligament laxity.[12] In this case, it may even be confused with rheumatoid arthritis and/or JIA. However, OI shows no erosion alterations on radiographs or morning stiffness. The alterations observed in the TMJ and extremities in the present case could be attributed exclusively to JIA.

TMJ involvement is rare in OI. Only 6% of a population of adults with the disease reported having severe TMJ disorders.[13] This finding in a patient with a presumptive diagnosis of hereditary collagen disorder warned u of the possibility of other diagnoses, and the investigation was performed retrospectively, until the diagnosis of JIA was made.

JIA is a differential diagnosis that should be considered in pediatric patients with spontaneous fractures.[14] However, not only it is a rare form of osteoporosis, but also it does not include blue sclera and is not influenced by family history, as in the case described here. OI itself can have a crippling arthropathy form,[15] but its clinical course is different from JIA and does not usually include elevated inflammatory markers or the presence of rheumatoid factor.

Both OI and JIA cause an increased risk of fractures, with different mechanisms. OI patients have spontaneous fractures due to the disease pathogenesis that leads to the bone matrix impairment. In JIA, fractures can occur due to increase in inflammatory cytokines, reduction of secondary bone mass, physical inactivity, or osteoporosis caused by corticosteroids. As low bone density is found in both diseases, bisphosphonates are successfully. The efficacy of treatment and follow-up can be measured by annual bone densitometry.[16]

The use of bisphosphonates in childhood is well established, particularly the use of pamidronate in OI and idiopathic juvenile osteoporosis.[17]

The authors would like to emphasize that this curious and rare association should be considered when investigating severe joint deformities, especially in the TMJ and extremities, associated with joint destruction in patients with a long-term picture of OI.

Conflicts of interest

The authors declare no conflicts of interest.

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